Case Report

Caudal regression syndrome: A rare case report with review of the literature

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ABSTRACT

Caudal regression syndrome is a very rare neural tube disorder, in which the caudal vertebral column and spinal cord develop abnormally, causing substantial sensory and motor deficits, primarily in the legs. An abnormality in the spinal cord and nerve roots interacts with a section of lumbar, lumbosacral, or coccygeal spinal dysgenesis or agenesis. Here, we present a rare case of caudal regression syndrome in a 3-year-old male child who was brought to the pediatrics department with a serious complaint of urine dribbling since infancy. The infant was discovered with spina bifida, lumbar scoliosis, and cavus deformity in both feet during a routine health check. For which, a neurological opinion was taken. Plain X-ray revealed a partial sacral agenesis, MRI revealed anterior and posterior nerve roots of cauda equina divided into two bundles of fibers due to abnormal path. The lower back and both gluteal areas were underdeveloped. On the basis of radiological findings, a diagnosis of the syndrome was confirmed.

Keywords: Caudal regression syndrome, Magnetic resonance imaging, Maternal diabetes, Radiodiagnosis

In caudal regression syndrome, the spinal cord and vertebral column are unable to grow in a usual manner due to a neural tube deficiency. Coccygeal dysgenesis (or agenesis) and abnormalities of the spinal cord and nerve root coexist in the same person. This condition is caused by the development of the faulty mesoderm [1]. The illness causes lower-limb neurological abnormalities such as bladder and bowel dysfunction as well as severe sensory and motor impairments, affecting an estimated 1:25,000 live births. Some teratogens have been reported to be involved in its development, including maternal diabetes, genetic variables, and environmental exposures [2,3]. An antenatal ultrasound is generally the first step in diagnosing caudal regression syndrome; however, an antenatal or postnatal magnetic resonance imaging (MRI) can be used to confirm the diagnosis.

CASE REPORT

A 3-year-old child was brought to the department of pediatrics with a major complaint of urine dribbling that had been persistent since infancy. The child was delivered naturally by vaginal delivery at full term and weighed 3.5 kg at birth. At the time of birth, the age of the mother was around 20's.

During a routine checkup, spina bifida, lumbar scoliosis, and cavus deformity in both feet were discovered. For which, a neurologist was consulted and advised for imaging evaluation by the department of radiodiagnosis.

A conventional radiograph, a computed tomography (CT) scan, and an MRI of the spine were all taken. X-rays showed partial agenesis of the sacrum (Fig. 1a). Coccyx and sacral segments (S2 to S5) were not evident on CT (Fig. 1b-d). The spinal cord abruptly halted at the D12 level, with no conus medullaris to be found, MRI revealed that the diameter of the central canal's lower dorsal section was increased. Nerve roots from both sides of the cauda equina produced a double bundle shape due to an abnormal path. The lower back and both gluteal areas were weak. Caudal regression syndrome was diagnosed based on imaging results (Fig. 2).

The parents were counseled about multiple surgeries required in view of genitourinary neuromuscular complications with urological and orthopedic inputs and were referred to a higher center for further treatment. After that, the patient was lost to follow-up.

DISCUSSION

The caudal vertebral column and spinal cord abnormality, known as sacral agenesis, are extremely uncommon and frequently random. It is expected to occur about once in every 25,000 normal pregnancies. In about 1 in 350, the risk is further increased due to the presence of chronic diseases such as diabetes mellitus. A female preponderance is seen with a male: female ratio of 2.7:1 [1,4].

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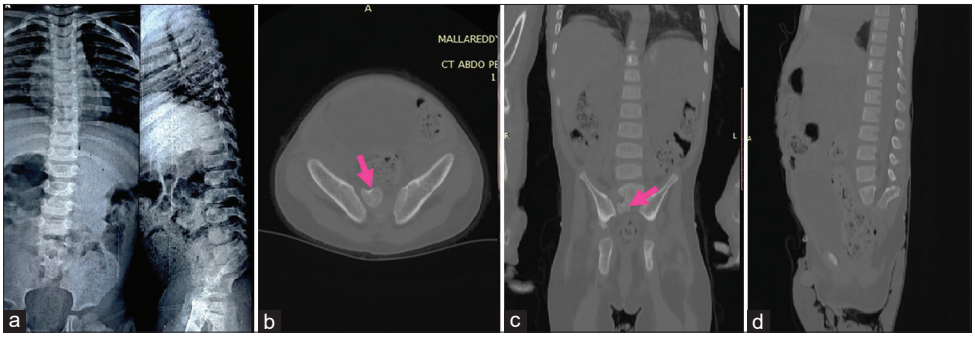

Figure 1: X-ray anteroposterior and lateral view. (a) Reveal partial sacral agenesis; (b) Axial; (c) coronal; and (d) sagittal non-contrast CT scan of the lower back shows the S1 sacral segment (pink arrow), as well as the non-visible sacral segments (S2 to S5) and the coccyx

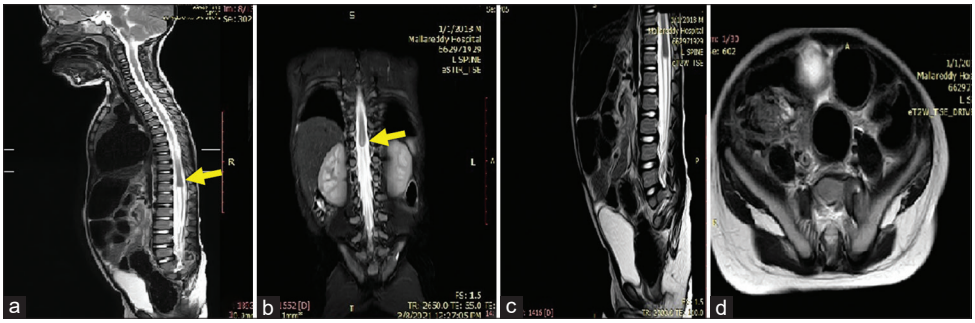

Figure 2: TSE T2W images in the sagittal (a and c), coronal (b), and axial (d) planes show an abruptly terminating spinal cord at D12 level with blunted appearance and absent conus medullaris (yellow arrow), mild dilatation of the central canal in the lower dorsal segment, abnormal cauda equina with separation of anterior and posterior nerve roots forming a double bundle shape, and hypoplastic muscle

The process of gastrulation takes place during the 3rd and 7th weeks of embryonic development [1]. Due to aberrant mesoderm development, sacral agenesis is possible. Damage to the mesoderm results in a loss of motor and neurologic function. Despite the fact that the cause has yet to be discovered, various explanations have been postulated, including a combination of genetic and environmental factors [4]. If a malfunctioning artery in the abdomen is to blame, it is possible that the defective mesoderm causes the artery to redirect blood away from the lower areas. Gestational hyperglycemia is certainly a teratogen, and there is compelling evidence that it contributes to the most severe form of caudal regression syndrome [2,3]. Genes and vascular under perfusion are also the other possible risk factors. Retinoids like retinoic acid have been shown to raise blood sugar.

Small intergluteal clefts and weak gluteal muscles are frequently encountered in persons with sacral agenesis. They exhibit talipes abnormalities, narrow hips, and leg atrophy in the distal portion. Around 20% of patients have subcutaneous lesions such as skin-covered lipomeningoceles, terminal myelocystoceles, or confined dorsal myeloschisis. Caudal spinal abnormalities have been linked to a variety of gastrointestinal, musculoskeletal, and cardiothoracic problems. Urinary and bladder dysfunctions are frequently found in patients with caudal regression syndrome. Among the syndromic complexes, where sacrococcygeal spine agenesis may be present are anorexia, teratomas, meningoceles, and tracheoesophageal fistula [5]. A triad of currarino (partial sacral agenesis, anorectal abnormalities, and presacral masses, such as teratomas and meningoceles) is also found [6].

According to the severity of regression, imaging appearances may differ. Due to the wedged or cigar-shaped structure of the conus medullaris and the extreme canal constriction that occurs from an improperly formed lumbar vertebral body rostral to the final full vertebral body, a truncated and blunt spinal cord is the outcome (dysgenesis/hypogenesis). First-trimester diagnosis may be difficult due to inadequate ossification of the sacrum. There may be an absent or hypoplastic sacrum, hypoplastic lower extremities, and the presence of shield sign (opposed iliac bones in absence of sacral vertebrae) which is typically seen on an axial scan and ultrasound of the unborn child. With the conus ending above the anticipated level, the fetal extremities can be observed in a crossed-legged tailor configuration (sometimes even higher than L1). The fetal MRI revealed those details that cannot be seen with ultrasound [7]. As an indicator of canal stenosis, it is possible to notice a distinctive wedge-shaped cord terminal.

According to the Pang, the patients with caudal regression syndrome may be divided into two broad groups on the basis of their MRI imaging [8]. Group 1 includes the patients with dilated central canal or a cyst filled with cerebrospinal fluid that can be related to the lower end of the conus medullaris. This group of individuals has severe sacral deformities and a significant sacral deficiency that extends to the S1 vertebrae or above. Group 2 includes thickened and extended filum terminale or intraspinal lipoma that bonds the conus medullaris below the normal level. This group is more prone to experience severe neurological issues. Lipomyelomeningocele, meningocele, terminal myelocystocele, and sacrococcygeal teratoma are among the possible differential diagnoses [9,10].

Compared to the general population, diabetic moms had a much higher rate of caudal regression. Malformations linked to

even a small rise in HbA1C need strict blood sugar management for diabetic women who wish to get pregnant. Pregnant women who plan their pregnancies and regulate their sugar levels before conception can avoid these anomalies, resulting in less emotional and physical trauma for the patients [11]. It requires substantial urologic and orthopedic procedures in a tertiary center, although the mental health of the survivors is normal. The early newborn mortality is connected with severe kinds of congenital heart disease, kidney disease, and respiratory disease. The early detection of caudal regression syndrome can also lead to abortion [12]. Standard prenatal care is provided if the pregnancy continues.

CONCLUSION

The imaging results of caudal regression syndrome are described in this case report. Compared to the general population, the incidence of caudal regression among diabetes moms is tens of thousands of times larger. Prenatal diagnosis can be done with ultrasound and fetal MRI; however, MRI is the preferred imaging modality in adults. The severity of the spinal abnormalities and any concomitant symptoms define the prognosis for caudal regression syndrome. It requires substantial urologic and orthopedic procedures in a tertiary center, although the mental health of the survivors is normal. Avoiding problems and enhancing prognosis can only be achieved by early detection and treatment. It is possible to terminate the pregnancy early when caudal regression syndrome has been diagnosed. Standard prenatal care is provided if pregnancy persists.

Ethical approval and consent

The parents of the child provided written informed consent for imaging. A documented informed consent form was signed by the patient's parents.

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